

## Course guide

### 200650 - EPIGEN - Genetic Epidemiology

Last modified: 09/06/2023

**Unit in charge:** School of Mathematics and Statistics  
**Teaching unit:** 1004 - UB - (ENG)Universitat de Barcelona.

**Degree:** MASTER'S DEGREE IN STATISTICS AND OPERATIONS RESEARCH (Syllabus 2013). (Optional subject).

**Academic year:** 2023    **ECTS Credits:** 5.0    **Languages:** English

#### LECTURER

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**Coordinating lecturer:** GUILLEM CLOT RAZQUIN

**Others:** Segon quadrimestre:  
GUILLEM CLOT RAZQUIN - A  
CRISTINA LÓPEZ GONZÁLEZ - A

#### PRIOR SKILLS

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Knowledge of basic statistical inference and generalized linear regression models.  
Basic knowledge of the use of the R analysis program.

#### DEGREE COMPETENCES TO WHICH THE SUBJECT CONTRIBUTES

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##### Specific:

MESIO-CE1. CE-1. Ability to design and manage the collection of information and coding, handling, storing and processing it.  
MESIO-CE2. CE-2. Ability to master the proper terminology in a field that is necessary to apply statistical or operations research models and methods to solve real problems.  
MESIO-CE3. CE-3. Ability to formulate, analyze and validate models applicable to practical problems. Ability to select the method and / or statistical or operations research technique more appropriate to apply this model to the situation or problem.  
MESIO-CE4. CE-4. Ability to use different inference procedures to answer questions, identifying the properties of different estimation methods and their advantages and disadvantages, tailored to a specific situation and a specific context.  
MESIO-CE5. CE-5. Ability to formulate and solve real problems of decision-making in different application areas being able to choose the statistical method and the optimization algorithm more suitable in every occasion.  
Translate to english  
MESIO-CE6. CE-6. Ability to use appropriate software to perform the necessary calculations in solving a problem.  
MESIO-CE9. CE-9. Ability to implement statistical and operations research algorithms.

##### Transversal:

CT3. TEAMWORK: Being able to work in an interdisciplinary team, whether as a member or as a leader, with the aim of contributing to projects pragmatically and responsibly and making commitments in view of the resources that are available.

CT4. EFFECTIVE USE OF INFORMATION RESOURCES: Managing the acquisition, structuring, analysis and display of data and information in the chosen area of specialisation and critically assessing the results obtained.

CT5. FOREIGN LANGUAGE: Achieving a level of spoken and written proficiency in a foreign language, preferably English, that meets the needs of the profession and the labour market.

## TEACHING METHODOLOGY

There will be sessions where the main concepts of each topic will be explained, which will be illustrated with examples of real or simulated data. Additionally, the student will have material with which he will be able to complement the concepts treated in the theoretical classes.

## LEARNING OBJECTIVES OF THE SUBJECT

- Knowledge of the biological principles necessary to be able to choose the most appropriate analyzes and to be able to draw appropriate conclusions.
- Knowledge of statistical analysis techniques to investigate the relationships between genes and diseases.
- Knowledge of statistical analysis techniques to assess the influence of the environment and the gene-environment association.
- Knowledge of the different types of study and which statistical analysis techniques are appropriate for each one.
- Treatment of data with appropriate structure to be used depending on the type of study.
- Knowledge of statistical methods of analysis of genetic data.
- Use of the necessary software to carry out the appropriate statistical analyzes

## STUDY LOAD

Type	Hours	Percentage
Hours large group	30,0	24.00
Self study	80,0	64.00
Hours small group	15,0	12.00

**Total learning time:** 125 h

## CONTENTS

### 1. Introduction to genetic epidemiology

#### Description:

Molecular genetics. Mendel's laws of inheritance. Modes of inheritance. Genetic map distances. Hardy-Weinberg equilibrium. Linkage disequilibrium

#### Full-or-part-time: 31h

Theory classes: 7h 27m

Practical classes: 3h 43m

Self study : 19h 50m

### 2. Classic studies

#### Description:

Familial aggregation studies. Segregation studies. Linkage studies. Heritability studies.

#### Full-or-part-time: 27h

Theory classes: 6h 29m

Practical classes: 3h 14m

Self study : 17h 17m

### 3. Genetic association studies

**Description:**

Association in family designs. Association in unrelated subject designs. Association with haplotypes or multiple markers. Gene-environment interactions.

**Full-or-part-time:** 27h

Theory classes: 6h 29m

Practical classes: 3h 14m

Self study : 17h 17m

### 4. Genome-wide association studies (GWAS)

**Description:**

Quality control of GWAS data. Association in GWAS. Population substructure. Single nucleotide polymorphism imputation. Post-association tasks. Polygenic risk scores.

**Full-or-part-time:** 40h

Theory classes: 9h 36m

Practical classes: 4h 48m

Self study : 25h 36m

## GRADING SYSTEM

### Continuous assessment

At the end of each of the blocks that make up the subject, an in-person test will be carried out in which theoretical questions must be answered and data analysed. The tests will be scored between 0 and 10, and the average of these scores will be the mark of the continuous evaluation of the subject (NC). If the student makes less than 75% of the NC tests, the qualification of the subject will be that of not presented.

If students want to modify the NC qualification, an optional test will be scheduled at the end of the course that will include the entire syllabus. The test may contain theory questions and data analysis. Only students who are considered presented in the continuous evaluation may take this optional test. The qualification of this test (NR) will be from 0 to 10.

The final grade of the course will be:

- NC for students who have only done continuous assessment.
- NR for students who take the additional test at the end of the course.

### Single evaluation

Those students who want to benefit from the single assessment will have to notify the course coordinator during the first 15 school days of the course.

The single evaluation will consist of a synthesis test that will include the entire syllabus of the subject. The synthesis test will receive a score between 0 and 10 and will correspond to the final grade for the subject.

The course will be considered approved if the final grade is higher than 5.

## BIBLIOGRAPHY

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### Basic:

- Ziegler, Andreas; König, Inke R. A Statistical approach to genetic epidemiology : concepts and applications. Weinheim: Wiley, cop. 2006. ISBN 9783527312528.
- Teare, M. Dawn. Genetic epidemiology. New York: Springer, cop. 2011. ISBN 9781603274159.
- Foulkes, Andrea S. Applied statistical genetics with R: for population-based association studies [on line]. New York: Springer Verlag, cop. 2009 [ Consultation: 28/06/2023]. Available on : <https://web-s-ebshost-com.recursos.biblioteca.upc.edu/ehost/ebookviewer/ebook?sid=a3ed510f-0d3a-4845-8238-4482a0884429%40redis&vid=0&format=EB>. ISBN 9780387895536.
- Gondro, Cedric. Primer to analysis of genomic data using R [on line]. Cham: Springer, 2015 [Consultation: 28/06/2023]. Available on : <https://ebookcentral-proquest-com.recursos.biblioteca.upc.edu/lib/upcatalunya-ebooks/detail.action?pq-origsite=primo&docID=2097291>. ISBN 9783319144740.
- Laird, Nan M.; Lange, Christoph. The fundamentals of modern statistical genetics [on line]. New York: Springer, 2011 [Consultation: 28/06/2023]. Available on: <https://link-springer-com.recursos.biblioteca.upc.edu/book/10.1007/978-1-4419-7338-2>. ISBN 9781461427759.
- González, Juan R.; Cáceres, Alejandro. Omic association studies with R and Bioconductor. Boca Raton: Chapman and Hall/CRC, 2019. ISBN 9781138340565.